



DEPARTMENT OF THE AIR FORCE  
59TH MEDICAL WING (AETC)  
JOINT BASE SAN ANTONIO - LACKLAND TEXAS

21 APR 2016

MEMORANDUM FOR SGCEE

ATTN: CAPT COLIN GALLAGHER

FROM: 59 MDW/SGVU

SUBJECT: Professional Presentation Approval

1. Your paper, entitled **LPR5 Mutation Case Presentation** presented at/published to **Third Coast Retina, Dallas, TX 23 April 2016** with MDWI 41-108, and has been assigned local file #**16148**.
2. Pertinent biographic information (name of author(s), title, etc.) has been entered into our computer file. Please advise us (by phone or mail) that your presentation was given. At that time, we will need the date (month, day and year) along with the location of your presentation. It is important to update this information so that we can provide quality support for you, your department, and the Medical Center commander. This information is used to document the scholarly activities of our professional staff and students, which is an essential component of Wilford Hall Ambulatory Surgical Center (WHASC) internship and residency programs.
3. Please know that if you are a Graduate Health Sciences Education student and your department has told you they cannot fund your publication, the 59th Clinical Research Division may pay for your basic journal publishing charges (to include costs for tables and black and white photos). We cannot pay for reprints. If you are 59 MDW staff member, we can forward your request for funds to the designated wing POC.
4. Congratulations, and thank you for your efforts and time. Your contributions are vital to the medical mission. We look forward to assisting you in your future publication/presentation efforts.

LINDA STEEL-GOODWIN, Col, USAF, BSC  
Director, Clinical Investigations & Research Support

PROCESSING OF PROFESSIONAL MEDICAL RESEARCH/TECHNICAL PUBLICATIONS/PRESENTATIONS			
1. TO: CLINICAL RESEARCH		2. FROM: (Author's Name, Rank, Grade, Office Symbol) Colin Gallagher, Capt, O-3, SGCEE	
3. GME/GHSE STUDENT: <input checked="" type="checkbox"/> YES <input type="checkbox"/> NO		4. PROTOCOL NUMBER: Case Report	
5. PROTOCOL TITLE: (NOTE: For each new release of medical research or technical information as a publication/presentation, a new 59 MDW Form 3039 must be submitted for review and approval.) Case Report			
6. TITLE OF MATERIAL TO BE PUBLISHED OR PRESENTED: LPR5 Mutation Case Presentation			
7. FUNDING RECEIVED FOR THIS STUDY? <input type="checkbox"/> YES <input checked="" type="checkbox"/> NO FUNDING SOURCE:			
8. DO YOU NEED FUNDING SUPPORT FOR PUBLICATION PURPOSES: <input checked="" type="checkbox"/> YES <input checked="" type="checkbox"/> NO			
9. IS THIS MATERIAL CLASSIFIED? <input type="checkbox"/> YES <input checked="" type="checkbox"/> NO			
10. IS THIS MATERIAL SUBJECT TO ANY LEGAL RESTRICTIONS FOR PUBLICATION OR PRESENTATION THROUGH A COLLABORATIVE RESEARCH AND DEVELOPMENT AGREEMENT (CRADA), MATERIAL TRANSFER AGREEMENT (MTA), INTELLECTUAL PROPERTY RIGHTS AGREEMENT ETC.? <input type="checkbox"/> YES <input checked="" type="checkbox"/> NO NOTE: If the answer is YES then attach a copy of the Agreement to the Publications/Presentations Request Form.			
11. MATERIAL IS FOR: <input checked="" type="checkbox"/> DOMESTIC RELEASE <input type="checkbox"/> FOREIGN RELEASE CHECK APPROPRIATE BOX OR BOXES FOR APPROVAL WITH THIS REQUEST. ATTACH COPY OF MATERIAL TO BE PUBLISHED/PRESENTED.			
<input type="checkbox"/> 11a. PUBLICATION/JOURNAL (List intended publication/journal.)			
<input type="checkbox"/> 11b. PUBLISHED ABSTRACT (List intended journal.)			
<input type="checkbox"/> 11c. POSTER (To be demonstrated at meeting: name of meeting, city, state, and date of meeting.)			
<input type="checkbox"/> 11d. PLATFORM PRESENTATION (At civilian institutions: name of meeting, state, and date of meeting.) Third Coast Retina, Dallas, Tx, on 23 April 2016			
<input type="checkbox"/> 11e. OTHER (Describe: name of meeting, city, state, and date of meeting.)			
12. EXPECTED DATE WHEN YOU WILL NEED THE CRD TO SUBMIT YOUR CLEARED PRESENTATION/PUBLICATION TO DTIC NOTE: All publications/presentations are required to be placed in the Defense Technical Information Center (DTIC).			
DATE April 22, 2016			
13. 59 MDW PRIMARY POINT OF CONTACT (Last Name, First Name, M.I., email) Silva, Debbie, deborah.silva@us.af.mil		14. DUTY PHONE/PAGER NUMBER 210-292-6573	
15. AUTHORSHIP AND CO-AUTHOR(S) List in the order they will appear in the manuscript.			
LAST NAME, FIRST NAME AND M.I.		GRADE/RANK	SQUADRON/GROUP/OFFICE SYMBOL
a. Primary/Corresponding Author Gallagher, Colin		O3	59 Trng Squadron/59MDG/SGCEE
b.			
c.			
d.			
e.			
f.			
I CERTIFY ANY HUMAN OR ANIMAL RESEARCH RELATED STUDIES WERE APPROVED AND PERFORMED IN STRICT ACCORDANCE WITH 32 CFR 219, AFMAN 40-401_IP, AND 59 MDWI 41-108. I HAVE READ THE FINAL VERSION OF THE ATTACHED MATERIAL AND CERTIFY THAT IT IS AN ACCURATE MANUSCRIPT FOR PUBLICATION AND/OR PRESENTATION.			
16. AUTHOR'S PRINTED NAME, RANK, GRADE Colin Gallagher, Capt, O3		17. AUTHOR'S SIGNATURE GALLAGHER.COLIN.FRANCIS.11 71271262 Digitally signed by GALLAGHER COLIN FRANCIS 11 Date: 2016.03.26 14:55:40 -0500	
18. DATE March 26, 2016			
19. APPROVING AUTHORITY'S PRINTED NAME, RANK, TITLE Walter Steigleman, CDR, Ophthalmology Program Director		20. APPROVING AUTHORITY'S SIGNATURE STEIGLEMAN.WALTER.A.118593 0137 Digitally signed by STEIGLEMAN WALTER A 118593 Date: 2016.03.28 09:41:41 -0500	
21. DATE March 28, 2016			



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<b>1st ENDORSEMENT (59 MDW/SGVU Use Only)</b>		
TO: Clinical Research Division 59 MDW/CRD Contact 292-7141 for email instructions.	22. DATE RECEIVED 3/30/2016	23. ASSIGNED PROCESSING REQUEST FILE NUMBER 16148
24. DATE REVIEWED 6 Apr 2016		25. DATE FORWARDED TO 502 ISG/JAC
26. AUTHOR CONTACTED FOR RECOMMENDED OR NECESSARY CHANGES: <input checked="" type="checkbox"/> NO <input type="checkbox"/> YES If yes, give date. _____ <input type="checkbox"/> N/A		
27. COMMENTS <input checked="" type="checkbox"/> APPROVED <input type="checkbox"/> DISAPPROVED The presentation is approved.		
28. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER Rocky Calcote, PhD, Clinical Research Administrator	29. REVIEWER SIGNATURE CALCOTE.ROCKY.D.1178245844 <small>Digitally signed by CALCOTE ROCKY D 1178245844              DN: cn=US, o=US, ou=Department, email=rocky.d@hhs.gov, c=US              Date: 2016.04.06 13:08:57 -0500</small>	30. DATE
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<b>3rd ENDORSEMENT (59 MDW/PA Use Only)</b>		
37. DATE RECEIVED 19 Apr 2016	38. DATE FORWARDED TO 59 MDW/SGVU 21 Apr 2016	
39. COMMENTS <input checked="" type="checkbox"/> APPROVED (In compliance with security and policy review directives.) <input type="checkbox"/> DISAPPROVED		
40. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER Joe Bela, GS-12, Director, Public Affairs	41. REVIEWER SIGNATURE BELA.JOE.JR.1130810990 <small>Digitally signed by BELA JOE JR 1130810990              DN: cn=US, o=US, ou=Department, email=joe.bela@hhs.gov, c=US              Date: 2016.04.21 10:16:40 -0500</small>	42. DATE 21 Apr 2016
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43. DATE RECEIVED	44. SENIOR AUTHOR NOTIFIED BY PHONE OF APPROVAL OR DISAPPROVAL <input type="checkbox"/> YES <input type="checkbox"/> NO <input type="checkbox"/> COULD NOT BE REACHED <input type="checkbox"/> LEFT MESSAGE	
45. COMMENTS <input type="checkbox"/> APPROVED <input type="checkbox"/> DISAPPROVED		
46. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER	47. REVIEWER SIGNATURE	48. DATE

PROCESSING OF PROFESSIONAL MEDICAL RESEARCH/TECHNICAL PUBLICATIONS/PRESENTATIONS		
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26. AUTHOR CONTACTED FOR RECOMMENDED OR NECESSARY CHANGES: <input checked="" type="checkbox"/> NO <input type="checkbox"/> YES If yes, give date. <input type="checkbox"/> N/A		
27. COMMENTS <input checked="" type="checkbox"/> APPROVED <input type="checkbox"/> DISAPPROVED The presentation is approved.		
28. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER Rocky Calcote, PhD, Clinical Research Administrator	29. REVIEWER SIGNATURE CALCOTE, ROCKY, D. 1178245844 <small>Digitally signed by ROCKY, D. 1178245844 DN: cn=Rocky, o=59 MDW, ou=59 MDW, email=rocky.d@59mdw.mil</small>	30. DATE
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31. DATE RECEIVED	32. DATE FORWARDED TO 59 MDW/PA	
33. COMMENTS <input checked="" type="checkbox"/> APPROVED (In compliance with security and policy review directives.) <input type="checkbox"/> DISAPPROVED Slide presentation includes the disclaimer required by the Joint Ethics Regulation. There are no ethics issues with making this presentation at the Third Coast Retina meeting on 23 April 2016.		
34. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER Arlene R. Christilles, GS-14, Chief, Civil Law	35. REVIEWER SIGNATURE <i>Arlene R. Christilles</i>	36. DATE 7 Apr 2016
<b>3rd ENDORSEMENT (59 MDW/PA Use Only)</b>		
37. DATE RECEIVED	38. DATE FORWARDED TO 59 MDW/SGVU	
39. COMMENTS <input type="checkbox"/> APPROVED (In compliance with security and policy review directives.) <input type="checkbox"/> DISAPPROVED		
40. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER	41. REVIEWER SIGNATURE	42. DATE
<b>4th ENDORSEMENT (59 MDW/SGVU Use Only)</b>		
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45. COMMENTS <input type="checkbox"/> APPROVED <input type="checkbox"/> DISAPPROVED		
46. PRINTED NAME, RANK/GRADE, TITLE OF REVIEWER	47. REVIEWER SIGNATURE	48. DATE

# HOW OLD?

COLIN GALLAGHER



## DISCLOSURES

The views expressed are those of myself, Dr. Gallagher, and do not reflect the official views or policy of the Department of Defense or its Components.

## HISTORY OF PRESENT ILLNESS

- Chief Complaint: Decreased red reflex of right eye
- HPI: 3 day old M referred from pediatrics for decreased leukocoria in right eye

## HPI CONTINUED

### PMHx:

- Term from G2P2
- Uncomplicated SVD
- Denied consanguinity
- (-) infectious screen
- Passed hearing screen

Schx: Non-contributory

Meds: None

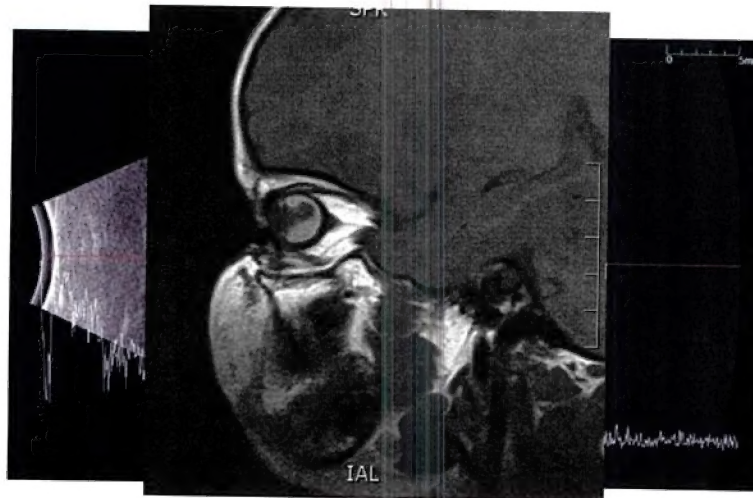
ALL: None

### PSHx:

- None



## FHX - SISTER AT 4 MO



## PHYSICAL EXAM

VA:

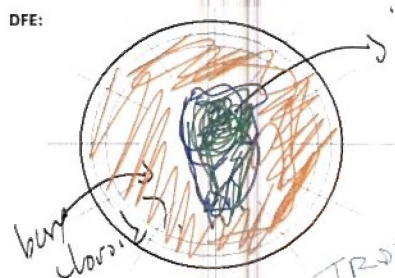
- OD: NBTL
- OS: BTL

External Exam:  
microphthalmic right eye

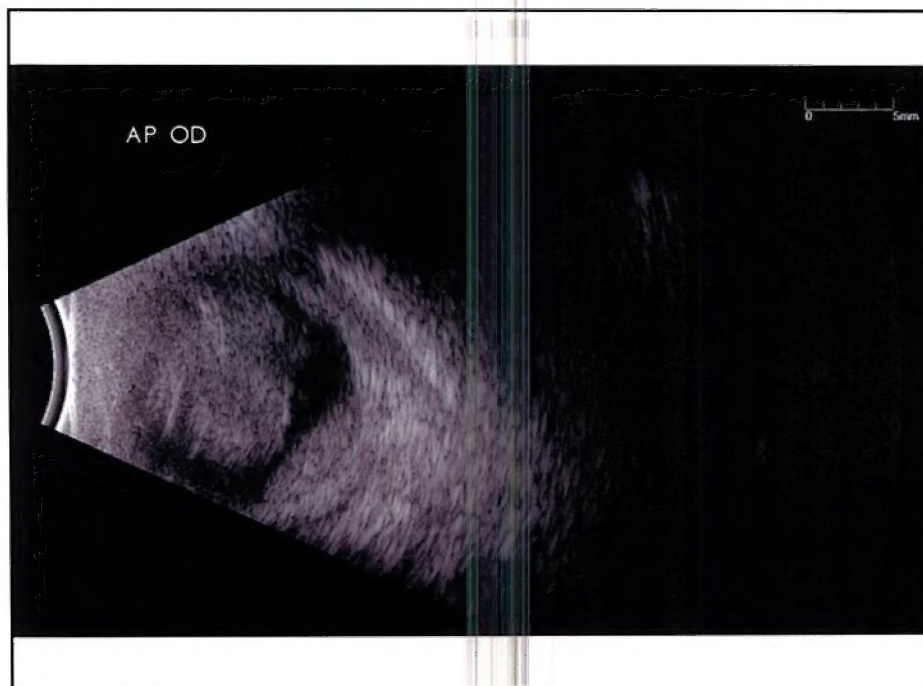
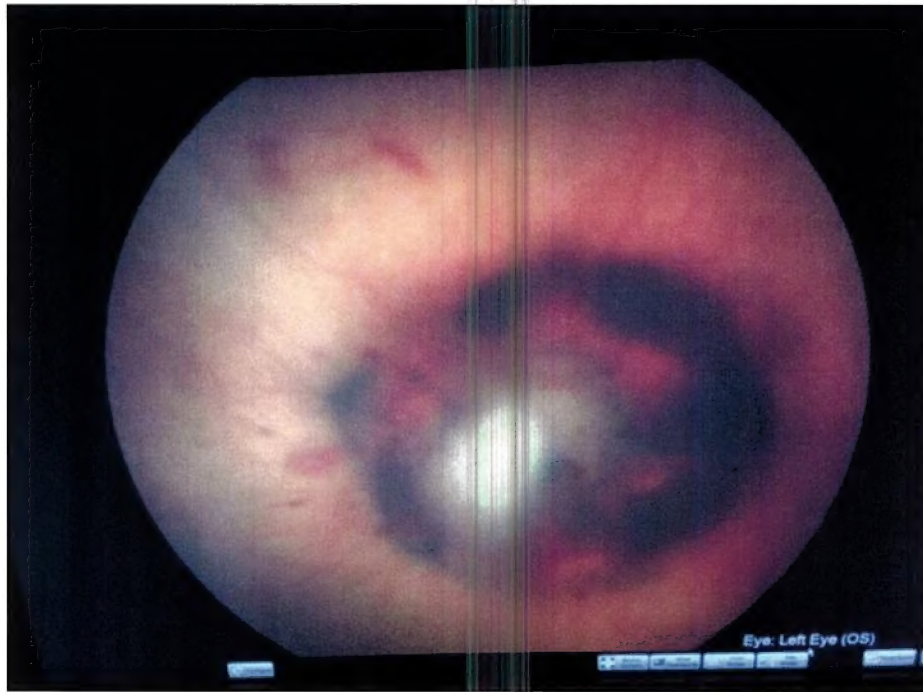
## PSLE:

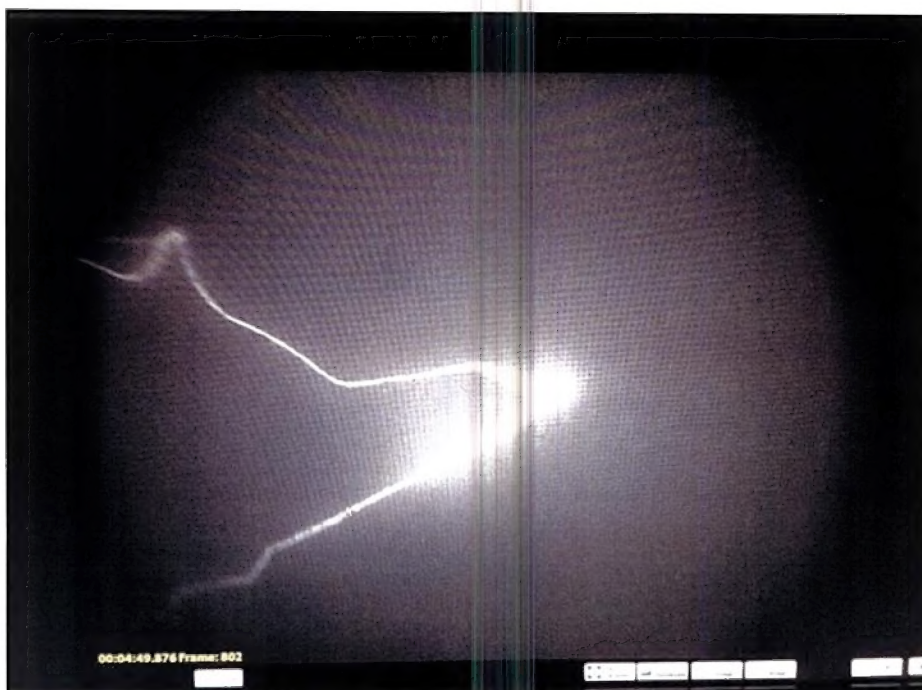
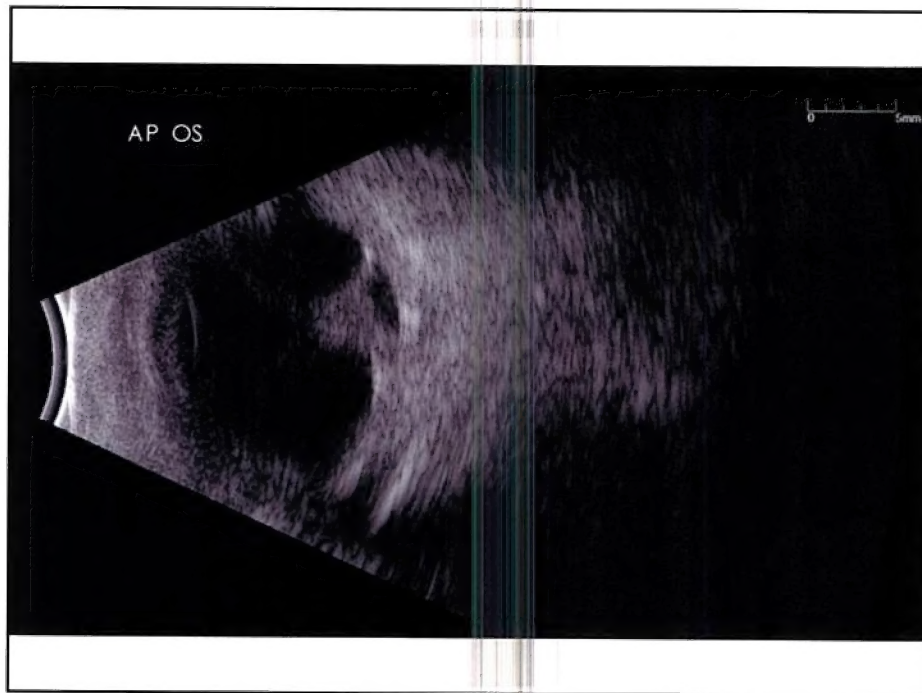
- LLL: Nml OU
- C/S: W & Q OU
- K: Clr & Q OU
- AC: D & Q OU
- I: F & R OU
- L: **retrolental opacity OD>OS**

## DILATED FUNDUS EXAM









## LABS

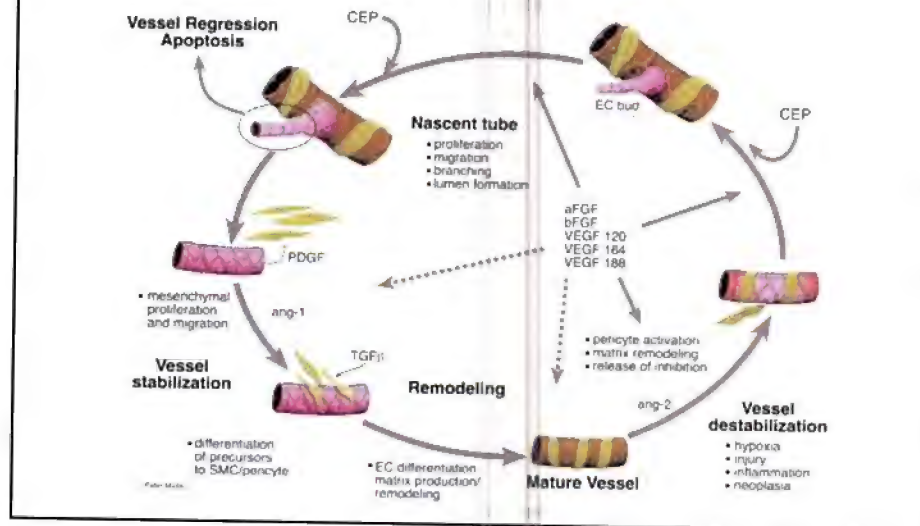
Test	Source	Result
Acylcarnitine Panel	Blood	Normal
Amino Acids Profile Neonatal	Blood	Normal
G6PD Gene Mutation Analysis	Blood	Normal
17-Hydroxyprogesterone	Blood	Normal
Cystic Fibrosis Screen	Blood	Normal
Biotinidase Neonatal	Blood	Normal
Thyrotropin Neonatal	Blood	Normal
Hemoglobinopathy	Blood	Normal
Galactose Neonatal	Blood	Normal

## DIFFERENTIAL DIAGNOSIS

- Retinoblastoma
- Retinopathy of prematurity
- Coats disease
- Norrie disease
- X-linked/AD familial exudative vitreoretinopathy
- Persistent fetal vasculature/persistent hyperplastic primary vitreous syndrome
- ATOH7 mutation



## FETAL VASCULATURE



## LITERATURE

### Pathophysiology and Mechanisms of Severe Retinopathy of Prematurity

by Elizabeth Hornett, MD

STUDIES ON THE PATHOGENESIS OF AVASCULAR RETINA AND NEOVASCULARIZATION INTO THE VITREOUS IN PERIPHERAL SEVERE RETINOPATHY OF PREMATURITY (AN AMERICAN OPHTHALMOLOGICAL SOCIETY THESIS)

by Mary Elizabeth Hornett MD

## LITERATURE

### Locus for Autosomal Recessive Nonsyndromic Persistent Hyperplastic Primary Vitreous

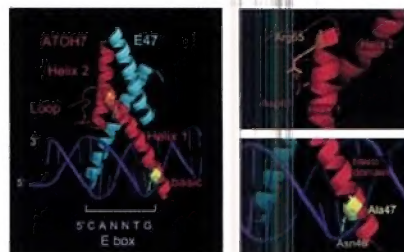
Shagufta Khaliq, Abdul Hameed, Muhammad Ismail, Khalid Anwar, Bart Leroy, Annette M. Payne, Shomi S. Bhattacharya, S. Qasim Mehdi

## LITERATURE

*Human Molecular Genetics*, 2012, Vol. 21, No. 16 3681–3694  
doi:10.1093/hmg/dds197  
Advance Access published on May 29, 2012

### ATOH7 mutations cause autosomal recessive persistent hyperplasia of the primary vitreous

Lev Prasov<sup>1,2</sup>, Tehmina Masud<sup>1,2</sup>, Shagufta Khaliq<sup>3</sup>, S. Qasim Mehdi<sup>4</sup>, Aiysha Abid<sup>4</sup>, Edward R. Oliver<sup>5</sup>, Eduardo D. Silva<sup>6</sup>, Amy Lewanda<sup>7</sup>, Michael C. Brodsky<sup>8</sup>, Mark Borchert<sup>9</sup>, Daniel Kelberman<sup>10</sup>, Jane C. Sowden<sup>10</sup>, Mehul T. Dattani<sup>11</sup> and Tom Glaser<sup>1,2,\*</sup>



## LITERATURE

### **Visual Acuity Outcomes with and without Surgery in Patients with Persistent Fetal Vasculature**

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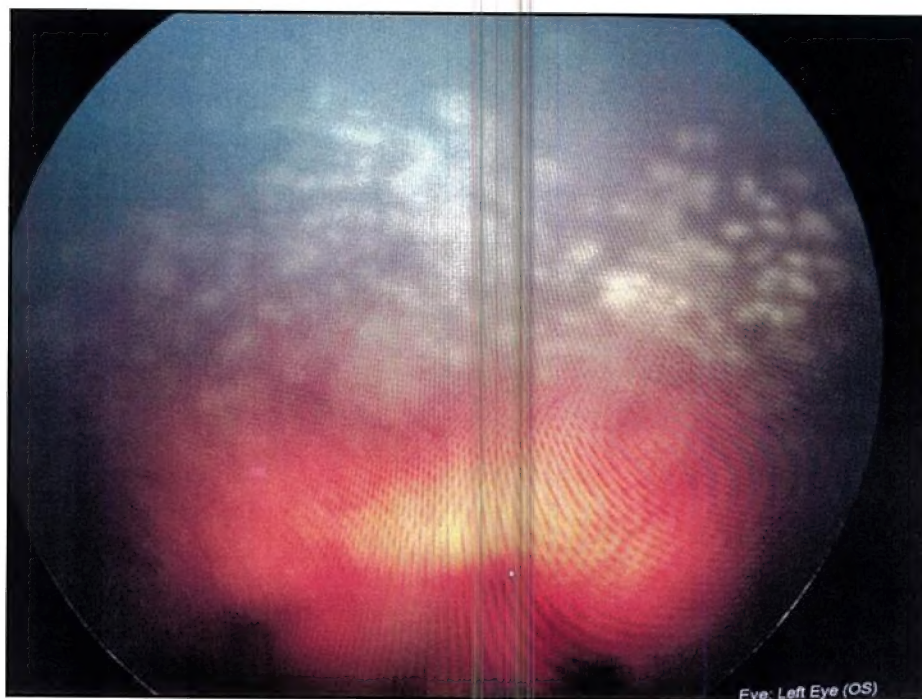
*George Alexandrakas, MD, Ingrid U. Scott, MD, MPH, Harry W. Flynn, Jr., MD, Timothy G. Murray, MD, William J. Feuer, MS*

## LITERATURE

### **Infant Aphakia Treatment Study: Effects of persistent fetal vasculature on outcome at 1 year of age**

*David G. Morrison, MD,<sup>a</sup> M. Edward Wilson, MD,<sup>b</sup> Rupal H. Trivedi, MD, MSCR,<sup>b</sup> Scott R. Lambert, MD,<sup>c</sup> and Michael J. Lynn, MS,<sup>c</sup> for the Infant Aphakia Treatment Study Group<sup>a</sup>*





## DISCUSSION

- Treatment options?
- Similar presentations?

## THANK YOU

Dr. Kim  
Dr. Grant  
Dr. Harper

## REFERENCES

1. Persistent Fetal Vasculature. In: Basic and clinical science course (BCSC) Section 6: Pediatric Ophthalmology and Strabismus. San Francisco, CA: American Academy of Ophthalmology; 2015:285-287.
2. Vitreous Development. In: Basic and clinical science course (BCSC) Section 2: Fundamentals and Principles of Ophthalmology. San Francisco, CA: American Academy of Ophthalmology; 2015:123.
3. Saint-Geniez and D' Amore, Development and pathology of the hyaloid, choroidal, and retinal vasculature, *Int. J. Dev. Biol.* 48: 1045-1058).
4. Prasov et al., ATOH7 Mutations cause autosomal recessive persistent hyperplasia of the primary vitreous, *Human Molecular Genetics*. 21(16):3681-3694
5. Alexandrakis et al., Visual acuity outcomes with and without surgery in patients with persistent fetal vasculature, *Ophthalmology*. (2000):1068-1072.
6. Morrison et al., Infant Aphakia Treatment Study: Effects of persistent fetal vasculature on outcome at 1 year of age, *Journal of AAPOS*. 15(5):427-431.
7. Walsh et al., Early vitrectomy effective for bilateral combined anterior and posterior persistent fetal vasculature syndrome, *Journal of Retinal and Vitreous Diseases*. 30(4): S1-S8.
8. Ober et al., Autosomal dominant exudative vitreoretinopathy, *British Journal of Ophthalmology*. 1980 (64): 112-120.
9. Harnett, M., Studies on the pathogenesis of avascular retina and neovascularization into the vitreous in peripheral severe retinopathy of prematurity (An American Ophthalmological Society Thesis), *Trans Am Ophthalmol Soc.* (2010) 108: 96-117.
10. Harnett M, Pathophysiology and mechanisms of severe retinopathy of prematurity. *Translation Science Review*. 2015; 122:200-210.